
Ethical dilemmas in clinical genetics

I D Young *Department of Child Health, Leicester Royal Infirmary, Leicester*

Author's abstract

This paper discusses the results of a survey of medical and paramedical opinion relating to various difficult ethical issues in clinical genetics. These include the confidentiality of the doctor-patient relationship, prenatal diagnosis and termination, and Huntington's chorea. It is suggested that this method provides a useful means of assessing what is ethically acceptable in contemporary society.

Introduction

There can be few areas of medicine which generate more controversy and debate than clinical genetics, a subject which has recently been discussed in this journal (1). Looked at simplistically, genetic counselling involves the straightforward provision of information to those who seek it, and at this level appears both harmless and worthy. In reality it can be much more complex. Situations arise which can stretch the confidentiality of the doctor-patient relationship to the limit, when for example a confirmed diagnosis is seen to have genetic implications for unsuspecting relatives. The provision of a risk estimate may, and in all probability will, be accompanied by guidance on how this risk may be reduced or side-stepped so that options such as antenatal diagnosis and abortion are raised.

All doctors must find themselves faced with difficult ethical decisions and wonder where to turn for guidance. For some the answer may lie in the acceptance of religious dogma. Others may feel that ethical values are more a matter for contemporary discussion and that the views of caring intelligent citizens may be sought in formulating acceptable standards. It is with this in mind that the author has sampled the opinions of medical, nursing and student staff on various practical problems.

Key words

Genetic counselling; prenatal diagnosis; Huntington's chorea; doctor-patient relationship.

Discussion

There is a very real danger in a survey of this nature that opinion will be readily forthcoming from those with extremist views, whilst the silent majority remains silent. Thus a vociferous minority may have a disproportionate impact. The replies in this study are drawn from a broad spectrum of experience and religious background and, although the response rate of 32 percent is disappointing, the diversity of opinion expressed suggests that it represents a reasonable cross-section of informed, interested medical or paramedical personnel. Whilst it is possible that the skewed age distribution may mean that idealism rather than pragmatism forms the basis of many of the replies, this may not necessarily be undesirable when trying to formulate a code of ethics.

The points raised in these questions can be considered under four headings. In questions 1-3 the doctor-patient relationship is under scrutiny. In almost every branch of medicine this relationship is sacrosanct. But with hereditary disease a confirmed diagnosis frequently has serious implications, not only for the patient but also for relatives, spouse, future children and perhaps also for society. How and whether such information should be processed, stored and disseminated to those at risk poses very real difficulties for the clinical geneticist. It is curious that 63 per cent of respondents feel that the employer of a dangerous worker should be informed against the patient's wishes, but only 29 per cent would approach at-risk relatives without permission. *The Handbook of Medical Ethics*, published by the British Medical Association (2) states that 'the importance of such information (to the relatives) probably outweighs the importance of complete individual medical confidentiality', a viewpoint which might leave many doctors uneasy and would almost certainly deter some patients from attending a genetics clinic even if they were aware of its existence.

Questions 4-9 relate to the particularly difficult subject of antenatal diagnosis and abortion. The areas of conflict which can arise in consideration of the 'rights' of the pregnant mother, the abnormal fetus (to live or to be aborted) and their doctor have recently been aired in a stirring commentary (3). It seems that

Questionnaire: methods and results

The questionnaire consisted of 12 questions. Respondents were asked to answer 'yes' or 'no' and to add comments if desired. Personal details including age, sex and religion, but not identity, were sought. Brief explanatory notes about the relevant diseases were included as in the appendix.

446 questionnaires were distributed at seminars, lectures and clinical meetings. 144 replies were received from 87 students, 40 nurses and 17 doctors. Their age distribution was:

	Male	Female	Total
15-19 years	14	21	35
20-24 „	27	28	55
25-34 „	10	16	26
35+ „	9	19	28
	60	84	144

The stated religious affiliations of the respondents were:

Protestant	- 54	Jewish	- 4
Roman Catholic	- 24	Hindu	- 3
Christian	- 7	Moslem	- 2
Atheist	- 34	Buddhist	- 1

No answer - 15

The questions and their responses were as follows. Answers have only been included which indicated a definite positive or negative response.

Question 1: Following the birth of a malformed mentally retarded infant with abnormal chromosomes, it is found that one of the parents has a balanced chromosomal rearrangement which led to the infant's problems. Other family members may also have this rearrangement and may therefore be at risk of having similarly affected children.

a) Are the child's parents justified in withholding this knowledge from their at risk relatives?

Answer: Yes - 13 No - 129

b) Should the doctor respect and act in accordance with their decision?

Answer: Yes - 94 No - 42

Question 2: Should unsuspecting individuals at risk of developing Huntington's chorea, such as someone who is adopted at birth and whose biological parent later develops the condition, be actively traced and informed of their risk?

Answer: Yes - 96 No - 35

Question 3: An adult presents with early signs of a slowly progressive hereditary disorder known to impair both intellect and co-ordination. This patient has a responsible position in which irresponsible behaviour could endanger other people's lives. Should the doctor inform the patient's employer against the patient's wishes?

Answer: Yes - 91 No - 44

Question 4: Is termination of pregnancy ever justifiable on the grounds of fetal abnormality?

Answer: Yes - 116 No - 16

Question 5: Is termination acceptable when it is known that the fetus has:

a) Down's syndrome? Yes - 95 No - 41

b) Spina bifida? Yes - 108 No - 32

c) Turner's syndrome? Yes - 49 No - 84

Question 6:

a) Is termination when the fetus is at 25 per cent risk of developing Huntington's chorea in later life acceptable?

Answer: Yes - 71 No - 67

b) Should termination only be offered in this situation if the lady will also agree to sterilisation?

Answer: Yes - 18 No - 114 (several cries of blackmail!)

Question 7: Is termination acceptable when the fetus has a condition which will not affect intellect but will cause severe physical handicap?

Answer: Yes - 85 No - 50

Question 8: Is termination acceptable when the fetus will be normal physically but will be mentally retarded?

Answer: Yes - 94 No - 34

Question 9: If you have answered 'yes' to any of questions 4-8, would you personally wish to have a termination in any of these circumstances, or if appropriate, encourage your spouse to do so?

Answer: Yes - 89 No - 21

Question 10: Should individuals who seek directive advice (for example 'but what would you do doctor?') be given it?

Answer: Yes - 71 No - 68

Question 11: Is directive counselling (for example 'I don't think you should have any children') ever justified?

Answer: Yes - 85 No - 59

Question 12: Is the medical profession justified in pursuing pre-clinical tests for Huntington's chorea?

Answer: Yes - 116 No - 10

litigation in the United States is directed at the obstetrician who fails to inform or abort, whilst in this country there are those who would wish to see quite the opposite situation.

These problems are particularly pertinent when the condition in question is either relatively mild, such as Turner's syndrome (which may be detected 'inadvertently' by amniocentesis for Down's syndrome), or is amenable to treatment, as in congenital adrenal hyperplasia, the prenatal diagnosis of which led to a lively correspondence a few years ago (4,5). Presumably there will be an even greater furor when disorders such as phenylketonuria become detectable prenatally by the application of molecular genetics, an event which is probably not far off (6). The majority views expressed in this survey suggest that termination is acceptable when the fetus is likely to be severely mentally or physically handicapped, but that there is considerable unease when this is not the case as in Turner's syndrome. Several respondents spontaneously commented that the ultimate decision should rest with the parents, an option which on the one hand might be seen as absolving the doctor of moral responsibility and on the other could be interpreted as consistent with a policy that 'when on the horns of a dilemma the operative word should be compassion' (7).

Questions 10 and 11 touch upon the topical issues of whether it is acceptable to offer advice, as opposed to information, to those who seek it and whether it might even be reasonable to thrust such advice upon patients (8). Both questions elicited roughly similar answers with a majority favouring a positive response to each. This is surprising since it is generally held that genetic counselling should be non-directive (1) and there is evidence that a more direct or paternalistic approach may be counter-productive or at best no more effective (as gauged by subsequent childbearing) than non-directive counselling (9). Perhaps the traditional image of the doctor as a kindly avuncular figure who 'knows best' still holds, and some patients do seem unable to reach a decision without direction (10). However, it could be argued that there is a thin line between eugenics and genetic counselling and that this line should be seen not to be crossed.

It is almost impossible to discuss ethics and genetics without mention of Huntington's chorea, a disorder in which counselling is fraught with difficulty. In question 2 a large majority felt that individuals at risk of developing this condition should be sought out and fully informed, suggesting that the right to information is paramount. Yet in question 3 a similar majority would be willing to risk this individual's livelihood when he or she developed the condition. The development of a predictive test which could reliably distinguish between those with and without the gene received almost universal approval in question 12, an attitude not shared by all members of the medical profession (11), although several surveys of opinion

within Huntington's chorea families suggest that most, but by no means all, individuals at high risk would welcome a reliable predictive test despite the absence of effective treatment (12, 13, 14).

Huntington's chorea is but one example of the many hereditary disorders which can raise grave ethical problems for the medical profession, problems which are unlikely to diminish and which will almost certainly increase in keeping with the rapid expansion of knowledge and understanding of genetic disease. It is suggested that the views of health care professionals, rather like the jury system, may provide a valuable means of assessing what is acceptable when formulating a practical code of ethics.

Appendix

EXPLANATORY NOTES AS INCLUDED WITH EACH QUESTIONNAIRE

In Down's syndrome, affected individuals have a characteristic appearance and almost invariably are significantly mentally retarded with an IQ ranging from approximately 25 to 55. About 20 per cent of Down's syndrome children die in infancy, whilst the remainder may live well into their fourth or fifth decade. It would be very unlikely that an individual with this condition could live an independent existence.

Girls with Turner's syndrome tend to be short (around 4' 6") and the majority have a slightly unusual, but by no means grossly abnormal, appearance. Intellect is usually normal. These patients are almost invariably infertile, and require long-term hormone therapy. This condition is occasionally encountered as an incidental finding when checking the chromosomes of a fetus for other conditions such as Down's syndrome.

The vast majority of children with spina bifida have major physical handicaps involving lower limb weakness or complete paralysis, in association with problems of bowel and bladder control. Such children may require frequent surgery in childhood for correction of orthopaedic problems and hydrocephalus. Older surviving children tend to have considerable emotional problems.

Huntington's chorea is a hereditary condition with age of onset roughly between 20 and 70 years, in which affected individuals show progressive mental and physical deterioration, leading to death within 10 to 20 years from onset. There is no effective cure. Each time an individual with this gene has a child there is a 1/2 chance that the child will inherit it.

References

- (1) Seller M J. Ethical aspects of genetic counselling. *Journal of medical ethics* 1982; 8: 185-188.
- (2) *The handbook of medical ethics*. London: British Medical Association, 1980: 30.

- (3) Milunsky A. Genetics, law and obstetric practice. *British journal of obstetrics and gynaecology* 1983; 90: 497-500.
- (4) Sibert J R. Antenatal detection of congenital adrenal hyperplasia. *Lancet* 1979; 2: 37.
- (5) Greenberg F. Prenatal diagnosis of congenital adrenal hyperplasia. *Lancet* 1979; 2: 908.
- (6) Smith T. How will the new genetics work? *British medical journal* 1983; 286: 1-2.
- (7) Smithells R W. *Ethical problems*. Archives of Disease in Childhood 1982; 57: 965.
- (8) Anonymous. Directive counselling (editorial). *Lancet* 1982; 2: 368-369.
- (9) Czeizel A, Metneki J, Osztovcics M. Evaluation of information - guidance genetic counselling. *Journal of medical genetics* 1981; 18: 91-98.
- (10) Leonard C O, Chase G A, Childs B. Genetic counselling - a consumer's view. *New England journal of medicine* 1972; 287: 433-439.
- (11) Thomas S. Ethics of a predictive test for Huntington's chorea. *British medical journal* 1982; 284: 1383-1385.
- (12) Stern R, Eldridge R. Attitudes of patients and their relatives to Huntington's disease. *Journal of medical genetics* 1975; 12: 217-223.
- (13) Teltscher B, Polgar S. Objective knowledge about Huntington's disease and attitudes towards predictive tests of persons at risk. *Journal of medical genetics* 1981; 18: 31-39.
- (14) Tyler A, Harper P S. Attitudes of subjects at risk and their relatives towards genetic counselling in Huntington's chorea. *Journal of medical genetics* 1983; 20: 179-188.